HEREDITY AS A CAUSE OF HUMAN BLINDNESS

A Review

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THE world-wide economic depression, the universal wave of crime, intensified nationalism, development of eugenics, and many other factors have combined at this time as never before to compel nations to face the problems of individual physical and mental handicap. Studies and surveys of criminality, insanity, deafness, blindness and other anomalies are receiving unheard-of-attention. Classics are being written in all these fields.

Hitler has decreed for the purification of German stocks so far as is possible by the sterilization of all persons known by the hereditary nature of their physical deformities to bear defective germ plasm. The subject is receiving attention in England. The Eugenics Research Association meeting this year was devoted principally to Race Integrity.

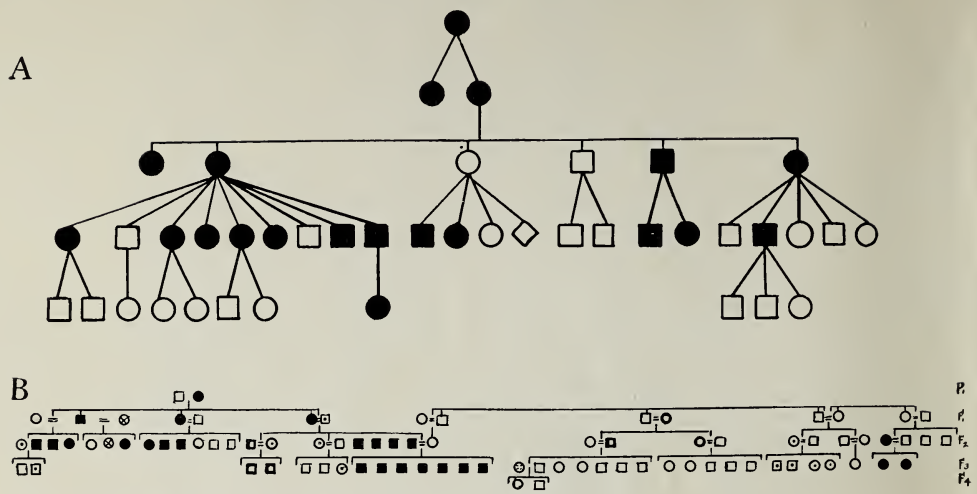
The most striking fact about the whole race-purification movement is the preponderant rôle which heredity plays in the production of all these physical and mental deformities which it is hoped may be eliminated. The radical says that the movement is timely; the conservative avers that we lack sufficient information about the nature of these handicaps upon which to base valid conclusions. But whether timely or premature the race-purification movement is upon us.

Blindness in America presents a peculiar problem. Here "Prevention of Blindness" has been for many years the slogan of much worthy propaganda, but thus far these efforts have failed to take cognizance of the enormous hereditary factor involved. It is true that in many individual cases it is impossible to differentiate between the defects produced by disease, faulty development, or heredity. This calls for intensive ophthalmological study and a sharpening of our diagnoses. It calls for investigations of individual pedigrees.

Information Inadequate

Our examination of institutional files reveals that while a few institutions maintain splendid records, the majority of them are in a very unsatisfactory condition and that conclusions based upon these records are inaccurate. Even where accurate diagnoses are made and records are perfectly kept, the deliberate falsehoods entered upon the records by relatives of the inmates destroy much of their value. The records of one State School for the Blind which we have examined show that a mother wrote upon the separate admission questionnaires for each of her four blind children that no person in the family save the applicant had ever been blind. She had a fifth blind child at home still under school age.

Our questionnaires sent to the various states show that the records kept of state and county expenditures for schools, sight-saving classes, aids, readers, printing, pensions, etc., are insufficient to evaluate accurately the many items necessary for computing the cost of blindness. The best statistics we may hope for are inaccurate estimates. With these almost insurmountable difficulties in mind, great credit is due to Dr. Harry Best for his brave attempt to collect statistics upon blindness.



INHERITANCE OF PIGMENTATION OF THE EYE

Figure 1

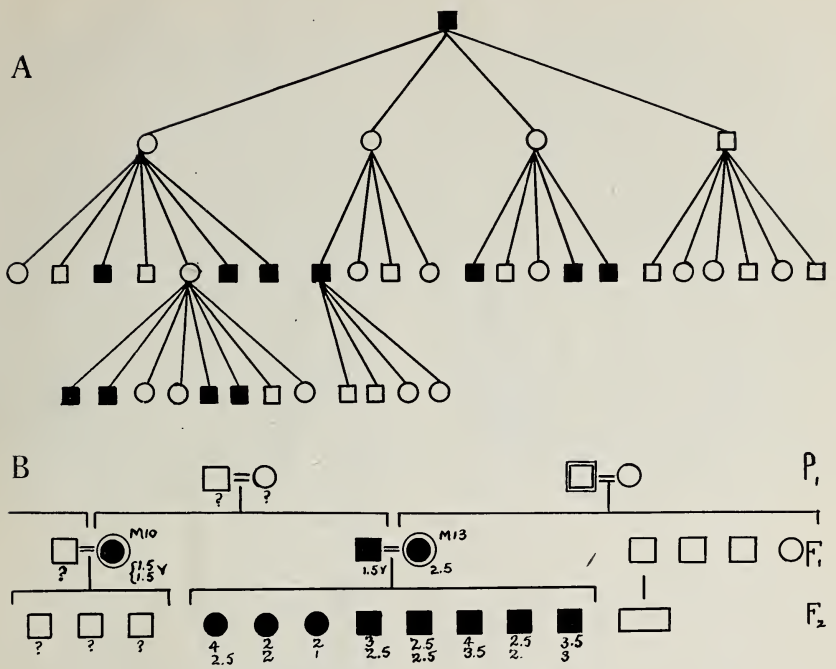
- *Blue with narrow, colored iris margin.*
- *Blue with light coloring matter about the periphery.*
- *Scattered pigment, especially a corona about the pupil in light eyes.*
- *Simplex (blue or gray).*
- *Dark brown (not completely diffuse).*
- *Scattered spots in lighter eyes.*

Pigmentation of various parts of the eye is a matter of vital importance to its proper function. The absence of pigment, as in true albinism, results in serious limitation of vision. The distribution of pigment in and around the eye is largely controlled by heredity. At (*A*) is given the pedigree of a family (after R. Peters) showing the inheritance for five generations of dark eyelids. At (*B*) is given a pedigree showing the inheritance of iris color for four generations, showing the tendency toward dominance of more intense pigmentation. From "Das Menschliche Auge."

Fifty-five chapters compose his recent volume on the subject.* The titles of some representative chapters are: Causes of Blindness and Extent of its Possible Prevention; Blindness and Heredity, Blindness and Disease; Blindness and Accidents; Organized Measures for Prevention of Blindness; The Blind by Sex, Age, Race, Nativity; Physical and Mental Condition of Blind; Education; Economic Condition; Cost of Blindness.

We are particularly interested in what Dr. Best has to say about heredity. "The proportion of the blind with blind relatives who have blind parents, in the aggregate (alone or in combination with other relatives) is 33.3 per cent; who have blind brothers or sisters in the aggregate, 71.2 per cent; and who have blind children in the aggregate, 6.1 per cent. The proportion with blind relatives who have blind parents alone is 24.1 per cent; who have blind brothers or sisters alone, 61.4 per cent; and who have blind children alone, 4.2 per cent. The proportion having one parent blind is 32.3 per cent, and that having both parents blind, 1.0 per cent. . . . The proportion who have both blind parents and blind brothers or sisters is 8.7 per cent." Best estimates 11.0 per cent of all blindness to be due to heredity! "Of those blind persons having a parent blind, practically three-tenths (31.3 per cent) have in addition brothers or sisters who are blind. This proportion is almost four times as great as the corresponding proportion for those not having a blind parent but having blind brothers or sisters (8.8 per cent)." We may suggest

*Best, Harry. *Blindness and the Blind in the United States.* Price $6.50. The Macmillan Company. New York, 1934.



CORNEAL DEFECTS INHERITED

Figure 2

(*A*) is a pedigree showing recessive sex-linked inheritance of keratosis follicularis, a form of degeneration of the cornea. Note that affected males transmit the character to their grandsons through their daughters (who themselves do not show the defect). The sons of affected males neither have the disease nor transmit it. Below at (*B*) is shown a pedigree of corneal astigmatism. The degree of astigmatism is shown under the symbols in diopters, which give a numerical expression of the extent to which the eye tends to be a cylinder rather than a sphere. Astigmatism may also be due to defects in the lens or to unequal muscular contraction.

that this difference is probably due in a large measure to dominant defects in the first case and recessive defects in the second. Dr. Best mentions Dr. Loeb's 1909 study. "In all the families in which blindness of hereditary character is recorded of the children born 60.7 per cent are blind. . . . In families where heredity is collateral—through cousins, or uncles or aunts—66.4 per cent of children are blind." We may suggest that many of these latter cases represent families where the parents are normal overlaps.

Best lists defects having hereditary tendencies as progressive myopia, retinitis pigmentosa, detachment of the retina, cataract, glaucoma, old age, trachoma, neuralgia, exposure to heat and sore eyes. He quotes Loeb's list which adds albinism, aniridia and coloboma iridis, anophthalmus, microphthalmus, atrophy of the optic nerve, ectopia lentis, familial corneal degeneration, megalophthalmus, nystagmus, ophthalmophlegia and ptosis (including strabismus). To these Best again adds others less common which may have hereditary influences: optic neuritis, macular and other retinal degeneration; nodular and reticular opacities of the cornea; syphilis in the form of interstitial keratitis; retinitis punctata albescens; distichiasis; buphthalmus; certain forms of choroiditis, certain forms of iritis; certain forms of glioma;

certain forms of amertropia; amaurosis from certain types of degeneration; blindness from idiocy; myopia and astigmatism. "Of the blind having parents who are first cousins, 37.5 per cent have blind relatives, as compared with 11.0 per cent for the blind in general." In respect to the value of eugenics to problems of blindness Best says: "So far as the increased possibility of blindness resulting from the marriage of blind persons is concerned, there is little need to fear, purely from the standpoint of eugenics, that the amount thereof will prove considerable. This is true even with respect to the marriage of the blind with the blind, though on other grounds strong objections are to be advanced to such unions." Evidently, Best sees the matter from the standpoint of increasing the number of hereditary blind over and above the number now existing, rather than from the standpoint of reducing as much as possible the present number of hereditary blind. Most geneticists will disagree with some of the views expressed in this chapter. The remaining chapters are filled with facts of paramount importance to the sociologist and worker for the blind. A great range of literature is cited. Even though we may not concur with Best's opinions upon heredity, we must admit that in its specialized field this volume will long stand alone.

The Genetics of Eye Defects

The hereditary types of defect producing blindness which Best shows to constitute at least 11.0 per cent of all blindness are admirably treated by Waardenburg in his 631 page volume, listing more than 120 types of hereditary ocular variation, and illustrated with 8 colored plates and 197 text-figures.* A sample of the completeness of this work is his treatment of the subject of albinism. Whereas most geneticists distinguish but one or two types of albinism, Waardenburg divides pigment variations into the following classes:

1. Complete albinism.
2. Incomplete albinism.
3. Localized eye and fundus albinism.
4. Aplasia, especially hypoplasia of the macula lutea.
5. Aplasia, especially hypoplasia of the macula and other portions of the retina with night blindness, restriction of visual field and strong secondary nystagmus.
6. Nystagmus with high grade myopia, and weak sight without weakening of the macula and albinism.
7. Primary nystagmus without underdevelopment of macula, without albinism or albinoid tendencies (irregular dominant).
8. Primary nystagmus without ocular indications (recessive).
9. Primary nystagmus without ocular indications (recessive-sex-linked).

The first chapter consists of an introduction, the second deals with pigment variation. The third takes up the lids and clefts describing variations in lashes, lid-folds, position, form and size of clefts, variations in conjunctiva and coloration of the lids. The inheritance of the latter is illustrated by a pedigree from R. Peters which we include as Figure 1*A*.

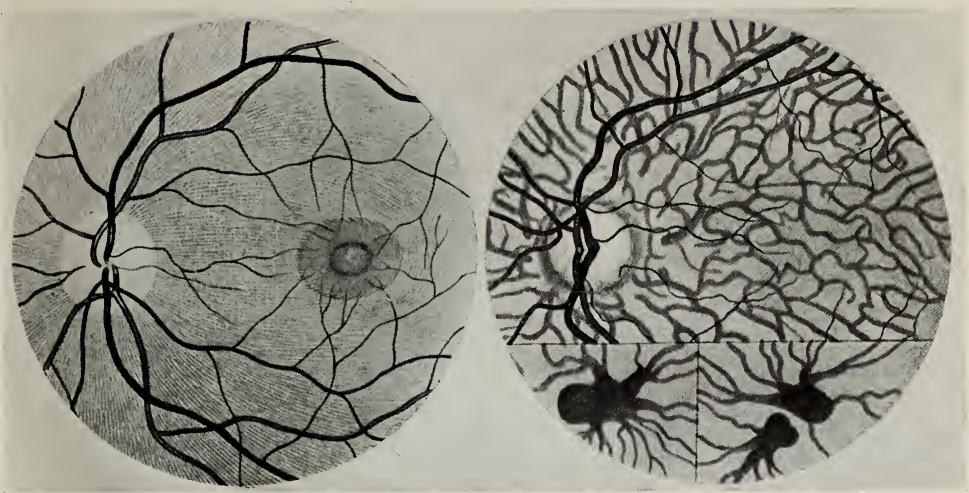
Under orbital variations are considered those of race and sex, individual variations, orbit and cleft, Dysostosis craniofacialis, dome heads, Dysostosis cleido-cranialis, exophthalmus, and facial asymmetry.

Waardenburg includes in the chapter on tear apparatus the tear glands, puncta, tear ducts, sac and naso-lacrimal ducts.

The subjects of ptosis, opththalmoplegia, paralysis, and abnormal movements of the lids are included in the chapter upon muscles.

The discussion of the cornea includes such interesting subjects as corneal astigmatism, keratocornus, corneal thickness, flat cornea, microcornea, cryptophthalmus, corneal degeneration and corneal pigmentation. In Figure 2*B* is shown a pedigree of astigmatism from Waardenburg and in Figure 2*A* one of corneal degenera-

*Waardenburg, P. J. *Das Menschliche Auge und seine Erbanlagen.* Price 30 gilders. Martinus Nijhoff, The Hague, 1932.



RETINA OF NORMAL AND ALBINO EYES

Figure 3

At the left is shown an enlarged view of the retina of a normal eye showing the region of the fundus and the veins and arteries which supply the retina. At the right is the same region of an albino eye. Note the remarkable system of blood vessels which radiate from the fundus of the albino eye which are invisible in the normal. From Waardenburg, *Das Menschliche Auge.*

tion from Waardenburg after Laméris-Rochat.

The variations of the sclera include the interesting subject of blue sclerotics. These individuals display a number of curious defects in addition to the blue eyeball,—brittle bones, deafness, thin sclera, degeneration of Bowman's and Decemet's membranes, oedema of the sclera, stunted growth, late teething and brittle teeth.

Hereditary vascular variations are many and include persistent pupillary membranes, size and form of pupil, ectopia pupillae, normal pigmentation, heterochromia, hypo- and hyper-pigmentations. Iris structure, hyperplasia of the iris layers, underdevelopment of iris muscles, multiple pupils, aniridia, coloboma of the iris, etc., are also discussed. In Figure 1*B* is shown a pedigree of eye color from Waardenburg.

The manifold forms of cataract are dealt with under the subject of lens as well as absence and underdevelopment of the lens, ectopia lentis, dislocation of the lens and correlated anomalies.

Short chapters consider the vitreous, zonule and pressure phenomena such as hydrophthalmus and glaucoma. A huge chapter covers variations of the optic nerve and the retina. Here one finds much information upon coloboma of the macula, hypoplasia and aplasia of the fovea and macula, total colorblindness, progressive degeneration of the macula, amaurotic family idiocy and night blindness. Also retinitis pigmentatosa, and other retinal degenerations, retinal tumors, atrophy of the optic nerve, and coloboma of the optic nerve are discussed.

The general topic of correlations between eye defects and those of the nervous system is considered under such topics as cerebro-spinal ataxia, migraine, progressive muscular atrophy, word blindness and the like.

There is a chapter on the heredity of refraction phenomena which deals with such topics as far sightedness and near sightedness; another dealing with general variability, asymmetry, and individual ocular characters. Another chapter has to do with hereditary ocular variations as manifest from the study of identical twins, while another considers the vision

of children born to related parents. The last chapter consists of a conclusion which is followed by comprehensive author and subject indices.

The nature of the colored illustrations but not their fine quality is indicated by the accompanying plates (reproduced in Figure 3) from the chapters on albinism. The rich variety of hereditary ocular variations dealt with we suggest with a series of pen drawings sketched from actual photos in Waardenburg's book (Frontispiece). Unfortunately, several halftones are printed too dark to be of much value, and the photographs of most patients are of a size too small to illustrate their defects satisfactorily for those not already familiar with them. The reader will note but probably not be confused by the several methods employed in the preparation of pedigree charts. There is an enormous bibliography.

No institution or individual interested in the subject of human blindness from any angle whatsoever can afford to be without these two classics.

The Value of Twin Research

To the Editor of The Journal of Heredity:

This letter is in response to the solicitation of opinion contained in Prof. H. H. Newman's article on identical twins reared apart, which appeared in the April, 1934, issue of the Journal of Heredity.

To Prof. Newman's question as to whether interest in such studies of identical twins reared apart, as he has made, is such as to warrant a continuation of his program, the answer, in my opinion, is unquestionably yes.

Not only he, but any other competent observer should put on record in the literature all cases of that kind which may come to attention and be accessible to investigation.

I believe it to be necessary to accumulate an amount of such material which would be sufficient for statistical treatment: at least one hundred cases.

I have a further suggestion to offer, namely, that a similar number of cases of identical twins reared together be investigated by precisely the same technique for use as control material. On the basis of observation of such cases in my own collection, I feel certain that such differences as have been found by Prof. Newman between identical twins reared apart are also to be found between identical twins reared together, though, perhaps, not with the same frequency.

In such cases it is often possible to demonstrate that the differences are due not to external environmental factors but partly to differences in intrauterine conditions or to factors at work at the time of birth.

It seems to me that only a comparison of the statistical findings of the group studied with those of a control group, selected as above, would make possible a definite conclusion as to the precise parts played, respectively, by germinal, intrauterine, natal, and postnatal factors on intelligence and other measurable traits.

For added control, from another angle, I believe there would be considerable advantage to be gained from an investigation, by the same technique, of a group of same-sex fraternal twins reared together.

Inasmuch as there is strong evidence to indicate that there are sex differences in intrauterine, infant, and early childhood mortality, in the vulnerability of cerebral tissues; and, as a result, in the incidence of both relative and absolute mental deficiency and other conditions definitely attributable to birth trauma, the materials gathered should be scrutinized in such a way as to reveal the findings separately for the two sexes.

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